This Guideline was published in 1999 by the American College of Medical Genetics under the auspices of a grant from the New York State Department of Health to the American College of Medical Genetics Foundation.

The American College of Medical Genetics has a web site at:
http://www.faseb.org/genetics/acmg

For further information about health rules, regulations and public health law in New York State, please consult the Department's web site at:
http://www.health.state.ny.us/nysdoh/phforum/phforum.htm.

This Guideline was sponsored by the New York State Department of Health for initial distribution within New York State. For this reason, you will note reference to the New York State Genetic Services Directory and laboratories licensed by the New York State Department of Health to perform testing on specimens originating in New York State.

This Guideline was produced at a series of meetings during 1997 and 1998; final edits were made in May 1999.

Table of Contents

Purpose

Major congenital malformations are common, are the leading cause of neonatal mortality, contribute substantially to chronic disease morbidity and have a high societal cost. Primary care providers who encounter newborns with malformations should be able to deal with their medical needs as well as the impact of malformations on the family. Accurate diagnosis is required for appropriate management and for counseling families about prognosis, treatment options, recurrence risks and resources for further information and support. The explosion of new information pertinent to etiology of birth defects makes it most important for the primary care provider to remain current in this area.

This Guideline describes critical components of the diagnosis and initial management of newborns with one or more congenital malformations for use by health care providers who care for newborn infants, irrespective of specialty orientation. It was developed in response to the perception that some newborns with malformations receive inadequate evaluation while others are inappropriately or excessively investigated. The orderly process described in the Guideline includes components which should be part of the evaluation of any newborn with one or more congenital anomalies, but does not specify an inflexible sequence, recognizing that evaluation involves repeated synthesis and modification of the process as information is gathered. Although many of the components of the Guideline reflect usual practice, some aspects require greater emphasis in this setting. For purposes of completeness, a comprehensive process is described. It is hoped that this Guideline will help health care providers to better evaluate these newborns, use limited resources wisely and make appropriate referrals for more specialized care.

The Guideline is intended to cover common situations and is not meant to be encyclopedic. Providers need to consider their personal level of comfort and expertise as well as their geographical practice setting when evaluating more complex cases. If significant components of the process cannot be accomplished in the primary care setting, referral should be made to a medical genetics specialist.

Guideline
Evaluation of the Newborn with Congenital Anomalies:

Components of Evaluation and Care

This outline describes essential components of evaluation and care, but does not specify an inflexible sequence. Details are discussed in the Executive Summary.

An algorithm shown in *Figure 1* demonstrates a method of evaluating the newborn with one or more congenital malformations.

A pocket memory aid for these components of care is shown in *Figure 2*.

History
- Prenatal
- Perinatal
- Family history

Physical Examination
- Assessment of gestational age
- Growth parameters and measurements
- Comprehensive examination

Differential Diagnosis
- Single malformation
- Multiple malformations due to syndrome
- Multiple malformations, etiology unknown

Diagnostic Evaluation
- Imaging
- Chromosome analysis
- Other genetic tests
- Consideration of referral to specialist(s)

Working Diagnosis/Counseling the Family
- Counseling Principles
- Supportive Setting
- Content

Patient record
- Documentation of positive and negative findings
- Diagnostic considerations
- Issues discussed in counseling
- Management plan

Ongoing care and case management
- Written reports
- Written materials for families
- Ongoing care and referrals as indicated

Dealing with Uncertainty

Major Sources of Further Information

Gorlin RJ, Cohen MM, Levin LS. Syndromes of the Head and Neck, Third Edition. Oxford University Press, New York, 1990.

Jones KL. Smith's Recognizable Patterns of Human Malformation, Fifth Edition. W.B. Saunders, Philadelphia, 1997.

McKusick VM. Mendelian Inheritance in Man, 11th Edition. Johns Hopkins University Press, Baltimore, 1994.

On-line Mendelian Inheritance in Man. <http://www3.ncbi.nlm.nih.gov/Omim/>

Figure 1. Algorithm: Evaluation of the Newborn with One or More Congenital Malformations

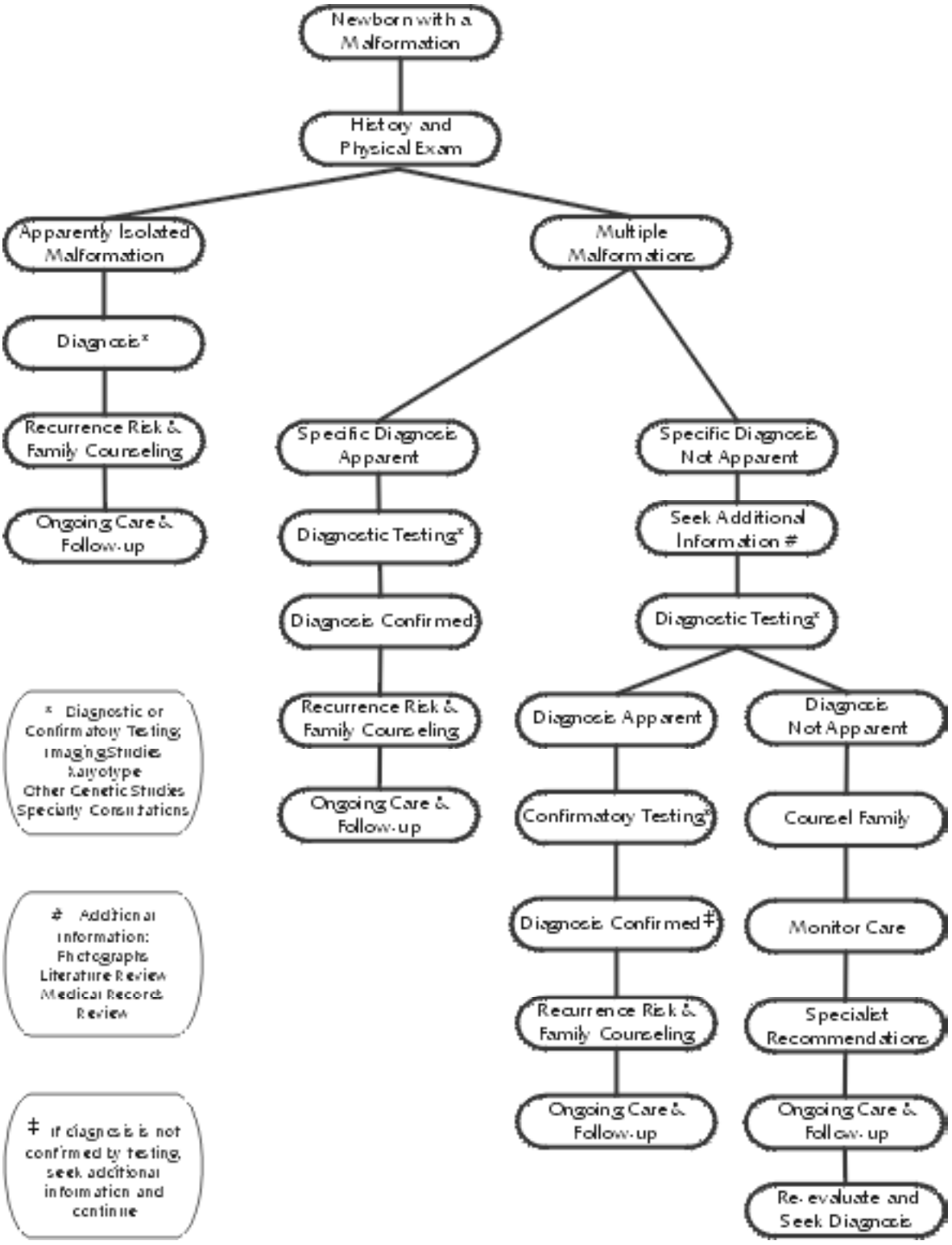


Figure 2: Components of Evaluation and Care Reference Card

Components of Evaluation and Care Reference Card
American College of Medical Genetics

Evaluation of the Newborn with Malformations

Components of Evaluation and Care

History

- Prenatal
  - Maternal age, parity, health
  - Complications of pregnancy
  - Illnesses and treatments
  - Exposures (drugs, etc)
  - Folic acid supplementation
  - Prenatal testing
- Perinatal
  - Duration of pregnancy
  - Intrapartum course
  - Presentation and mode of delivery
  - Complications and course
- Family history
  - Information re: three generations
  - Malformed infants
  - Stillborns
  - Familial disorders or traits
  - Consanguinity
  - Ethnic background

Physical Examination

- Gestational age, growth parameters and measurements (%iles)
- Comprehensive examination
  - General appearance
    - Proportionality and symmetry
    - Positioning, posture
    - Behavior
    - Other observations
  - Skin pigmentation, dimples, peeling, vascular or other lesions
  - Head shape, symmetry, fontanelles
  - Scalp hair patterning and whorls
  - Facial features
    - Pupils, orbits, palpebral fissure length and slant
    - Ear rotation, size and shape
    - Nasal appearance and patency
    - Lips, philtrum and vermilion border
    - Palate, tongue and alveolar ridges
    - Mandibular shape and size
  - Nuchal posterior hairline, redundant skin or webbing, sinus tracts
  - Chest shape, symmetry, location of nipples, +/- accessory nipples
  - Heart murmurs and pulses
  - Lung sound symmetry

Components of Evaluation and Care Reference Card
New York State Department of Health

Physical Examination (continued)

- Abdominal wall and palpation abnormalities, e.g. masses
- Genital appearance and maturation
- Anal orifice location and patency
- Back symmetry +/- sinus tracts or hair tufts in intergluteal cleft
- Extremity proportion, appearance, range of motion, abnormalities
  - Palmar creases, flexion creases at joints
- Neurological exam, including tone, alertness & reflexes

Initial Impression & Differential Diagnosis

- Single (isolated) malformation
- Multiple malformations of known pattern (syndrome identified)
- Multiple malformations (pattern not recognized)

Diagnostic Evaluation:

- Imaging
- Chromosomal analysis
- Other genetic tests
- Consider specialty referral and/or consultation

Develop Working Diagnosis and Counsel Family

Counseling the Family

- Counseling principles
  - Supportive
  - Respect privacy and confidentiality
  - Facilitate grieving process
- Supportive setting
  - Privacy and quiet
  - Inclusion of others according to family preference
- Content
  - Medical facts and diagnoses
  - Etiology and prognosis
  - Treatment plan and priorities
  - Uncertainties
  - Resources
  - Recurrence risks
  - Family psychosocial support

Longitudinal Care and Case Management

- Written reports and written materials for families
- Ongoing care and referrals as indicated

Note: This Executive Summary summarizes a more complete and fully referenced version which expands upon the points herein. Those who wish to obtain the full document, which includes the Guideline Justification, Appendices and References, should contact the New York State Genetic Services Program at 518-486-2215. The full document is also available from the NYS Department of Heath site on the internet: http://www.health.state.ny.us.

Executive Summary

Evaluation of the Newborn with Single or Multiple Congenital Anomalies: Statement of Purpose

Major congenital malformations are common, are the leading cause of neonatal mortality, contribute substantially to chronic disease morbidity, have a high societal cost and profoundly affect families.

This Guideline describes critical components of the approach to the diagnosis and initial management of newborn infants with one or more congenital malformations for use by health care practitioners who care for neonates, irrespective of specialty orientation. It was developed in response to the perception that some newborns with malformations receive inadequate evaluation while others are inappropriately or excessively investigated. The orderly process described in the Guideline includes components which should be part of the evaluation of any newborn with one or more congenital anomalies, but does not specify an inflexible sequence, recognizing that evaluation involves repeated synthesis and modification of the process as information is gathered. Although many of the components of the Guideline reflect usual practice, some aspects require greater emphasis in this setting. For purposes of completeness, a comprehensive process is described. Adherence is intended to facilitate an accurate diagnosis and appropriate family counseling. Expected positive outcomes include a more consistent approach to assessment, improved determination of prognosis, better management of affected newborns, improved cost-benefit and provision of accurate information to families. If this cannot be accomplished by the primary care provider, referral should be made to a specialist in medical genetics. Even with the most comprehensive workup, 30-60% of infants with malformations will not be given a specific diagnosis.

Sources for Referral and Consultation:

New York State Genetic Service Providers, *Appendix 4*
American College of Medical Genetics (301) 530-7127
National Society of Genetic Counselors (610) 872-7608, mail box #7
State Genetics Coordinators, *Appendix 5*
March of Dimes Birth Defects Foundation (914) 428-7100
New York State Genetic Services Program (518) 486-2215
Organization of Teratology Information Services (801) 328-2229
New York State Developmental Disabilities Council (800) 395-3372

These recommendations were developed by an interdisciplinary task force convened under the sponsorship of the New York State Department of Health and the American College of Medical Genetics Foundation. Participants included representatives from Family Medicine, Pediatrics, Obstetrics, Pediatric Surgery, Medical Genetics, Public Health and consumer groups (see *Table 1* for participating organizations). The Guideline is based on a review of the available literature and a consensus of expert opinion, with recognition that evidence-based literature in this area is limited. Because of its length, the entire Guideline (see Table of Contents) has not been distributed to all providers. Only the Table of Contents, Purpose, Algorithm,

Table 1. Organizations Represented on the Clinical Guidelines Team for Evaluation of the Newborn with Single or Multiple Congenital Anomalies*

Alliance of Genetic Support Groups
American Academy of Family Physicians
American Academy of Pediatrics, New York State Chapter
American College of Medical Genetics
American College of Obstetrics and Gynecology
American College of Physicians, New York State Chapter
American College of Surgeons
American Public Health Association, New York State Affiliate
Council of Regional Networks for Genetic Services
International Society of Nurses in Genetics
Links, Inc.
March of Dimes Birth Defects Foundation
National Society of Genetic Counselors
New York State Task Force on Life and the Law
Organization of Teratology Information Services
Society of Craniofacial Genetics

** see Appendix 8 for further information about each organization*

and Executive Summary have been mailed to most. References and specific citations do not appear in this short version, but are provided in the expanded Guideline Justification segment. Those wishing a more detailed elaboration of the statements and recommendations in the Executive Summary - with supporting references - should request the complete document from the New York State Department of Health's Genetic Services Program at (phone) 518-486-2215 or (fax) 518-473-1733. The complete version of this document is also available from the New York State Department of Health site on the Internet: http://www.health.state.ny.us.

Definitions. (Additional definitions are found in the Glossary, *Appendix 1*.)

Major congenital anomaly/malformation: a structural abnormality present at birth which has a significant effect on function or social acceptability; examples: ventricular septal defect, cleft lip

Minor congenital anomaly/malformation: a structural abnormality present at birth which has minimal effect on clinical function but may have a cosmetic impact; example: preauricular pit

Developmental variant/variation: a cosmetically and functionally insignificant structural deviation from the usual, of prenatal origin and usually familial; example: fifth finger clinodactyly

Components of Evaluation and Care.

When a newborn with one or more malformations is identified, a detailed history and physical examination must be undertaken to ascertain whether additional malformations are present and to seek a specific etiologic diagnosis. Diagnostic studies should be selected based on the information elicited and a working diagnosis should be developed. The family should receive detailed counseling in a setting and with content that is appropriate to their needs. Medical records and reports should reflect available laboratory and clinical data, diagnostic considerations and a plan for ongoing care, evaluation and management.

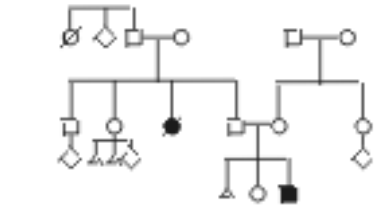
Although reaching an etiologic diagnosis for the newborn with multiple malformations is a primary goal of the evaluation process, a specific diagnosis might not be apparent after detailed evaluation and diagnostic testing. For a variety of reasons, such as age-dependent phenotypic or behavioral manifestations or uniqueness of the pattern of malformations, diagnosis is not always apparent in the newborn period.

History

A comprehensive history is a critical component of the evaluation of a newborn with single or multiple malformations. The sequence described suggests an orderly passage from history to physical examination to diagnostic evaluation. However, in actual practice, the history and physical examination represent a dynamic and interactive process in which certain elements of history may be sought after the physical exam. Likewise, certain physical parameters may be assessed further and more carefully based on information derived from historical data. Additionally, results of initial diagnostic testing may suggest the need for further history or physical examination. The following elements should be included:

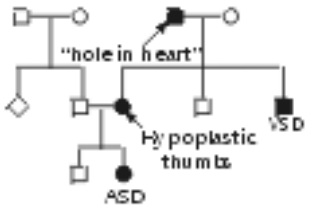
- Prenatal history
  - Maternal age, parity and health, including maternal illnesses and medications used
  - Onset and quality of fetal movements throughout pregnancy
  - Pregnancy complications
  - Viral and parasitic infections
  - Teratogenic exposures, such as alcohol, tobacco, drugs and medications
  - Periconceptional supplementation with folate
  - Prenatal testing (nature of testing and where performed)
- Perinatal history
  - Duration of pregnancy
  - Intrapartum course and duration
  - Intrapartum drug or medication exposure
  - Presentation and mode of delivery
  - Complications of delivery and infant's condition at birth (Apgar score)
  - Description of placenta
  - Birth weight; appropriate for gestational age? (See Physical Examination section)
  - Neonatal course, including diet, medications and complications
- Family history
  - A three generation family history (*see example pedigrees in sidebar*) with health information about all relatives, including parents, siblings, grandparents, uncles, aunts and cousins and noting any instances of reproductive losses or infertility
  - Specific information should be elicited regarding:
    - any relative with mental retardation or known malformations
    - infants in immediate or extended family with malformations or birth defects
    - neonatal deaths, stillbirths or childhood deaths in immediate or extended family

1. Down Syndrome due to Unbalanced Chromosome Translocation



● Down Syndrome
◐ Mentally retarded

2. Holt-Oram Syndrome



● Holt-Oram Syndrome

*See Glossary for key to symbols.

- familial disorders or physical features that "run in the family"
- Consanguinity in parents
- Ethnic background
- Prior genetic testing or screening

Medical records and correspondence regarding parents, siblings or other relatives should be reviewed to corroborate any significant positive findings elicited through the history.

Physical Examination

A complete physical examination must be performed, with particular attention to major and minor malformations and to physical variations. The same areas should be examined in other family members, when appropriate. Medical photography is invaluable, particularly to enable documentation of changes over time and for the purpose of referral. Essential components of the newborn physical examination include:

- Growth parameters
  - Assessment of gestational age by physical parameters (*see Appendix 2A*)
  - Length, weight and head circumference, with percentiles (*see Appendix 2B*)
  - Assessment of proportionality and symmetry
  - Specific measurements where indicated by observation, such as inner-canthal distance or upper to lower segment ratio (*see Appendices 2C-J*)
- General appearance
  - Tone, posture, positioning, alertness, vigor, color, respiratory effort and other observations
- Detailed examination
  - Skin - pigmentation pattern (areas of increased or decreased pigmentation), dimples, vascular or other lesions, or excessive peeling
  - Head - shape, symmetry, fontanelles
  - Scalp - hair patterning and location of hair whorls
  - Facial features
    - Eyes - pupils, orbits (hyper or hypotelorism) including palpebral fissure inclination and length
    - Ears - location, rotation, configuration and size, patency
    - Nose - appearance and patency of nares
      - appearance of nasal bridge and columella
    - Mouth - appearance of upper lip, philtrum and vermilion border
      - intra-oral examination of palate, alveolar ridges and tongue
    - Mandible - shape and symmetry
  - Neck - posterior hairline, presence of sinus tracts, torticollis, redundant skin or webbing
  - Chest - shape, symmetry, circumference, location of nipples, accessory nipples
  - Cardiovascular - heart murmurs, pulses, blood pressure
  - Lungs - symmetry of breath sounds
  - Abdomen - appearance of umbilicus, muscle tone, integrity of wall, enlarged organs or masses
  - Genitalia - size, appearance, palpation of testes (in males), presence of ambiguity
  - Anus - location and patency

- Back - symmetry, spine, presence of sinuses or hair tufts in inter-gluteal cleft
- Extremities - proportions, appearance, range of motion (including hips), pulses, presence of reduction or duplication of segments
- Hands and feet - nails; creases (palmar, phalangeal and flexion); joints
- Neurological - tone, response, alertness, reflexes

Initial Impression and Differential Diagnosis

The history and physical findings should lead to an initial impression and differential diagnosis. These will guide selection of preliminary tests, the content of initial counseling of the family and development of an immediate plan for management, which can be modified as new information is developed and synthesized. Initial impression should fit into one of three categories:

- Single (isolated) malformation
- Multiple malformations, recognizable pattern (syndrome identification)
- Multiple malformations, pattern not recognized

Diagnostic Evaluation

Diagnostic tests should be selected to clarify or establish a clinical diagnosis when possible. Such tests may be of particular value when a syndrome pattern is not recognized or to facilitate risk assessment for genetic counseling. Single (isolated) malformations or syndromes which are recognized on a clinical basis may not require additional diagnostic tests. It is important to discuss with the family the possible ramifications of genetic testing, including the implications for relatives.

- Select tests in a prioritized order, rather than using a "shotgun" approach; initial results can guide selection of more specific subsequent tests
- Consultations with specialists in pertinent fields may clarify diagnostic possibilities
- Request medical genetics consultation when a clear working diagnosis is not evident, to confirm a questionable diagnosis, to seek further information about an established diagnosis, or to provide detailed genetic counseling, particularly in complex situations.

  (See Appendix 4 for Genetic Service Providers in New York State and Appendix 5 for State Genetics Coordinators.)

Diagnostic tests which should be considered include:

- Diagnostic Imaging
  - Radiographs, computed tomography (CT), magnetic resonance imaging (MRI) or ultrasonography (US) should not be considered routine, but should be employed as indicated by the differential diagnosis.
- Chromosomal analysis

  Banded chromosomal analysis should be obtained in the following situations:
  - Infants with two or more major malformations
  - Infants with a single major malformation or multiple minor malformations who are also small-for-dates
  - Infants with a single major malformation who also have multiple minor anomalies

- In selected cases - usually in consultation with a medical geneticist - high resolution chromosome banding or fluorescence *in situ* hybridization (FISH) may be indicated to detect sub-microscopic structural chromosome changes or microdeletions (as in DiGeorge or Williams Syndrome).

A list of cytogenetics laboratories approved by the New York State Department of Health is included in *Appendix 6*. Updates are available on the web at *www.wadsworth.org/labcert/clep/clep.html*. Consideration should be given to compliance with federal and state regulatory requirements and professional accreditation, such as that provided by the American College of Medical Genetics or College of American Pathologists, when selecting a diagnostic cytogenetics laboratory.

◆ Other genetic tests

- Testing for the Fragile X syndrome is not generally useful in the evaluation of the newborn with single or multiple malformations, because congenital malformations are not a typical sign of the condition.
- Biochemical, metabolic or molecular genetic tests may be indicated in specific circumstances (*see Tables 2 and 3).* A list of genetic testing laboratories approved by the New York State Department of Health is included in *Appendix 7.* Updates are available on the web at *www.wadsworth.org/labcert/clep/clep.html*.

Table 2. Situations Suggesting the Need for Metabolic Testing in the Newborn with Malformations:

Selected clinical findings	Selected laboratory findings	Selected radiologic findings
Ambiguous genitalia	Metabolic acidosis	Punctate calcifications
Enlarged fontanelle	Abnormal liver function tests	Severe osteopenia
Seizures	Persistent hyperbilirubinemia	
Severe hypotonia	Hyperammonemia	
Cataracts	Hypocholesterolemia	
Coarse facies	Hypoglycemia	
Hepatosplenomegaly		
Lethargy or coma		
Persistent vomiting		
Unusual odor		

Working Diagnosis

The working diagnosis is established by data obtained from the history, physical examination and preliminary diagnostic results and forms the basis for counseling and longitudinal care. Positive findings in the family history should be confirmed by a review of medical records and/or family photographs when possible. Corroboration by collection and review of records may be required and is often invaluable in clarifying an otherwise confusing history.

Counseling the Family

The approach taken in counseling the family of a newborn with congenital anomalies sets the stage for future interactions. Counseling is an ongoing process; its staging and depth should be matched to each family. The items below should eventually be covered, but the depth of coverage in the initial session may vary according to family circumstances.

Table 3. A Sampling of Congenital Malformation Syndromes for which Specific Molecular Tests may be Available:

SYNDROME	MOLECULAR TEST
MICRODELETION SYNDROMES	
Williams	*ELN* (elastin)
Velocardiofacial/DiGeorge	Microdeletion within 22q11
Miller-Dieker	Microdeletion within 17p13.3
CRANIOSYNOSTOSIS SYNDROMES	
Apert and Crouzon	*FGFR2*
Pfeiffer	*FGFR1*
Saethre-Chotzen*	*Twist* or *FGFR3*
MALFORMATION SYNDROMES	
Treacher Collins	*Treacle*
Waardenburg*	*PAX3* or *HOX10*
Alagille	*Jagged*
BONE DYSPLASIAS	
Achondroplasia or thanatophoric dysplasia	*FGFR3*
Osteogenesis imperfecta*	*COL1A1* or *COL1A2*
Spondyloepiphyseal dysplasia congenita	*COL2A1*

** Linkage to two different genes has been found in different families*

- Counseling Principles
  - Supportive, family-centered, non-judgmental attitude
  - Respect for family privacy, confidentiality and autonomy
  - Sensitivity to ethno-cultural and language differences
  - Acknowledgment and facilitation of the grieving process
- Supportive Setting
  - Quiet, private location
  - Inclusion of additional family members, clergy, etc, as desired by family
- Content
  - Medical facts
  - Diagnostic considerations
  - Mechanisms and cause
  - Realistic prognosis and optimism when appropriate
  - Treatment plan and priorities
  - Acknowledgment of uncertainties
  - Early intervention and support resources
  - Recurrence risks and reproductive options
  - Assistance with family adjustment to information
  - Referral to psycho-social services as needed

Patient Record

In addition to information on the history and physical examination, the following should be included in the patient's file:

- Diagnostic considerations
- Documentation of positive and negative findings
- Management plan for further evaluation and treatment
- Issues discussed in counseling, including recurrence risk and availability of prenatal diagnosis including serial ultrasound monitoring of subsequent pregnancies

Privacy and confidentiality of the medical record must be assured.

Letters to families should include:

- Patient's name, diagnosis and means by which diagnosis was reached
- Brief summary of consultation
- Recurrence risk to relatives
- Availability of prenatal diagnostic testing
- Primary care provider's availability for ongoing discussion or referral
- Information on support groups and community resources

Where to find Genetic Support Groups:

Alliance of Genetic Support Groups
4301 Connecticut Avenue, NW, Suite 404
Washington, DC 20008-2304
(800) 336-GENE (4363)
e-mail: info@geneticalliance.org
<http:// www.geneticalliance.org/>

The National Organization for Rare Disorders, Inc.
P.O. Box 8923
New Fairfield, CT 06812-8923
(203) 746-6518 Fax (203) 746-6481
(800) 999-6673
<http://www.pcnet.com/~orphan>

March of Dimes Resource Center
1275 Mamaroneck Avenue
White Plains, NY 10605
(888) MODIMES (663-4637)
e-mail: resourcecenter@modimes.org
<http://www.modimes.org/rc/help.htm>

Longitudinal Care and Case Management

Newborns with one or more malformations should receive ongoing care and may require multidisciplinary care and case management. Some clinical problems or physical findings may evolve over time and become more apparent with age.

◆ Ongoing care of the newborn should be assumed by the primary care physician, who takes on the role of case manager, providing such care and making referrals as indicated.

◆ The primary care provider should be familiar with available resources such as community, educational and rehabilitation services.

◆ Management of complex conditions should include specialists at tertiary treatment centers.

◆ Families should receive relevant written information at an appropriate literacy level, translated if necessary, including some or all of the following:

- Summaries of medical facts, past testing, diagnoses and treatment plan
- Genetic counseling information
- Booklets or pamphlets about the diagnosis, if available
- Sources for further information and support, such as genetic disease support groups and community resources, if available.

Dealing with Uncertainty

A specific diagnosis may not be apparent after detailed evaluation and diagnostic testing. The following principles should be kept in mind when counseling a family:

◆ Inability to establish a specific diagnosis is not uncommon and is often perplexing and stressful for both families and health care providers. A specific diagnosis may not be reached for 30 - 60% of infants with malformations.

- Application of a poorly fitting diagnosis may do more harm than good.
- Innovative technologies are being developed and new diagnoses are being recognized in dysmorphology (the field concerned with malformations and physical variations) and genetics (*see Appendix 1. Glossary*).
- Longitudinal follow-up, re-examination and periodic literature review by the primary care provider and specialist are important aspects of ongoing care.
- Some diagnoses have age-dependent manifestations which may reveal a diagnosis that was not apparent at birth.
- Medical Genetic and/or other specialty consultation should be requested for unclear, rare or unknown diagnoses or for assistance with unusual diagnostic tests and complex counseling.
- Even in the most experienced hands, a diagnosis may not be reached.

Further Recommendations and Conclusions

The diagnosis and management of newborns with one or more congenital anomalies can be complex and at times requires coordination of multiple disciplines. The primary care provider should be aware of the following:

1. Every effort should be made to obtain an etiologic diagnosis in the newborn with one or more malformations. A specific diagnosis will enable the physician to understand the immediate and long term needs of the patient, mobilize the resources required to optimize outcome and provide information regarding education, genetic recurrence risks and support to the family.

2. Assignment of a final diagnosis may require ongoing observation and the incorporation of newly developed techniques as new data become available. The medical geneticist may serve as an important resource under such circumstances.

3. Staging of communication and information according to the family's needs is essential.

4. Families should receive information about the diagnostic process and diagnosis in a manner that is linguistically appropriate and ethnically and culturally sensitive. This includes, when known, information on natural history, prognosis, genetic recurrence risks and available resources. Such information should be transmitted in person and whenever possible in written form as well.

5. The primary care provider must be aware of some of the extensive resources available (see Appendices for examples) and use them when appropriate. In addition to the standard medical literature, this includes syndrome/birth defect compendia and databases, support group publications, information from public health agencies and other relevant sources specific to the condition.

6. Primary care providers should recognize their role in helping the family adjust to the impact of the birth of a child with a congenital disorder. Ongoing care and support are best provided by the primary health care provider, with assistance from the medical genetic specialist or other specialists when needed.

7. The primary care provider must ensure the confidentiality and privacy of genetic information.

General References and Resources

Books

American Academy of Pediatrics. Genetic Disorders and Birth Defects: A Compendium of Guidelines and Resources for the Primary Care Practitioner. American Academy of Pediatrics, Elk Grove Village, 1997.

Buyse M. Birth Defects Encyclopedia, 2 vols., Blackwell Scientific Publications, 1992.

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Zitelli BJ, Davis HW. Atlas of Pediatric Physical Diagnosis, Third Edition. CV Mosby, St. Louis, 1997.

On-Line Resources for Health Care Providers

Medical Search engines
- Medical Matrix <*http://www.slackinc.com/matrix/*>
- Cliniweb <*http://www.ohsu.edu/cliniweb/*>
- PEDINFO <*http://www.uab.edu.pedinfo/*>

On-line Mendelian Inheritance in Man (OMIM) (On-Line version of McKusick's book)
< *http://www3.ncbi.nlm.nih.gov/Omim/*>

Genetics Societies' Home Page
<*http://www.faseb.org/genetics.html*>

University of Kansas Genetics Professional Home Page (many links to other sources on genetics and congenital malformations)
<*http://www.kumc.edu/gec/geninfo.html*>

University of Kansas Support Groups Information and links
<*http://www.kumc.edu/gec/support/supgroup.html*>

Blazing a Genetic Trail (tutorial on basic genetics)
<*http://www.hhmi.org/GeneticTrail/*>

Description of Approach to the Dysmorphic Child
<*http://www.kumc.edu/instruction/medicine/pedcard/genetics/genetics.html*>

Pedinfo links for Congenital Diseases
<*http://www/UAB.EDU/pedinfo/DiseasesCongenital.html*>

List of Biochemical Genetic Tests by Disease (details of biochemical genetic testing)
<*http://biochemgen.ucsd.edu/wbgtests/dz-tst.htm*>

Helix Directory of Medical Genetics Laboratories (Password needed)
<*http://healthlinks.washington.edu/helix*>

Alliance of Genetic Support Groups
<*http://www.geneticalliance.org*>

March of Dimes Birth Defects Foundation
<*http://www.modimes.org*>

National Organization for Rare Disorders
<*http://www.pcnet.com/~orphan*>

March of Dimes Resource page
<*http://www.modimes.org/rc/help.htm*>

March of Dimes Fact Sheets On-Line/NOAH
<*http://www.noah.cuny.edu/pregnancy/pregnancy.html#BIRTHDEFECTS and gene*>

Genetic support group links
<*http://members.aol.com/dnacutter/sgroup.htm*>

Guide to genetic resources on the Web
<*http://www.dml.georgetown.edu/%7Edavidsol/len.html*>

Rare Genetic Diseases in Children
<*http://mcrcv2.med.nyu.edu/murphpo1/intro.htm*>